Figure 1-1

EXON (SIZE)	EXON 3'		INTRON (SIZE)		EXON 5'
EXON 1 (>460 bp)	GCT G	gtgagtagctccggc..	intron 1 (>30 kb)	..tttcctttattttag	CT TCG
	Ala A				la Ser
EXON 2 (171 bp)	CAA G	gcaagtgatactttc..	intron 2 (~2.3 kb)	..catattgatttatag	GA TAT
	Gln G				ly Cys
EXON 3 (171 bp)	CTC A	gtaagtaaagtaacc..	intron 3 (>30 kb)	..tttgttttcttttag	GT CCA
	Leu S				er Pro
EXON 4 (111 bp)	ACA G	gtaaaaattaccatt..	intron 4 (~3.8 kb)	..ttcctgttcttatag	GA GAC
	Thr G				ly Asp
EXON 5 (92 bp)	TTG GAG	gtaagtttgccgtta..	intron 5 (~6 kb)	..ttaaaacacttgcag	CTG ATT
	Leu Glu				Leu Ile
EXON 6 (231 bp)	CCC AAT	gtaagttcttcatag..	intron 6 (~4.1 kb)	..ttttcctctatatag	GGA TCT
	Pro Asn				Gly Ser
EXON 7 (115 bp)	GGA G	gtaagatagtcaata..	intron 7 (>7 kb)	..aaattatccaaacag	AT CAT
	Gly A				sp His
EXON 8 (161 bp)	AGC GAG	gtgagtgtatacaaa..	intron 8 (~1.6 kb)	..actctaatttatcag	GTT GGC
	Ser Glu				Val Gly
EXON 9 (148 bp)	CCA G	gtaaaaactactgtc..	intron 9 (>9.7 kb)	..tctacaaatccacag	GG GAA
	Pro G				ly Glu
EXON 10 (137 bp)	AGC CTG	gtaagaaaaaactaa..	intron 10 (>5 kb)	..tactttgtcttacag	GCA GTG
	Ser Leu				Ala Val
EXON 11 (173 bp)	GAA CG	gtaagaccctaaggg..	intron 11 (>20 kb)	..ctttctttctttaag	C AAC

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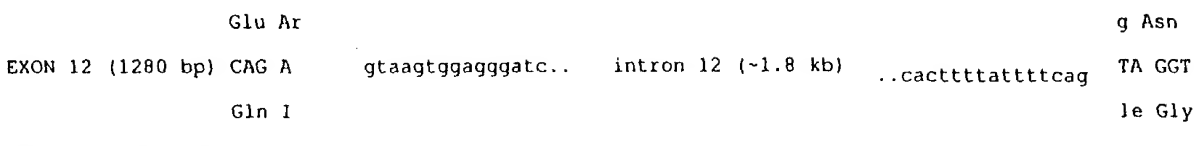
Figure 1-2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le Gly
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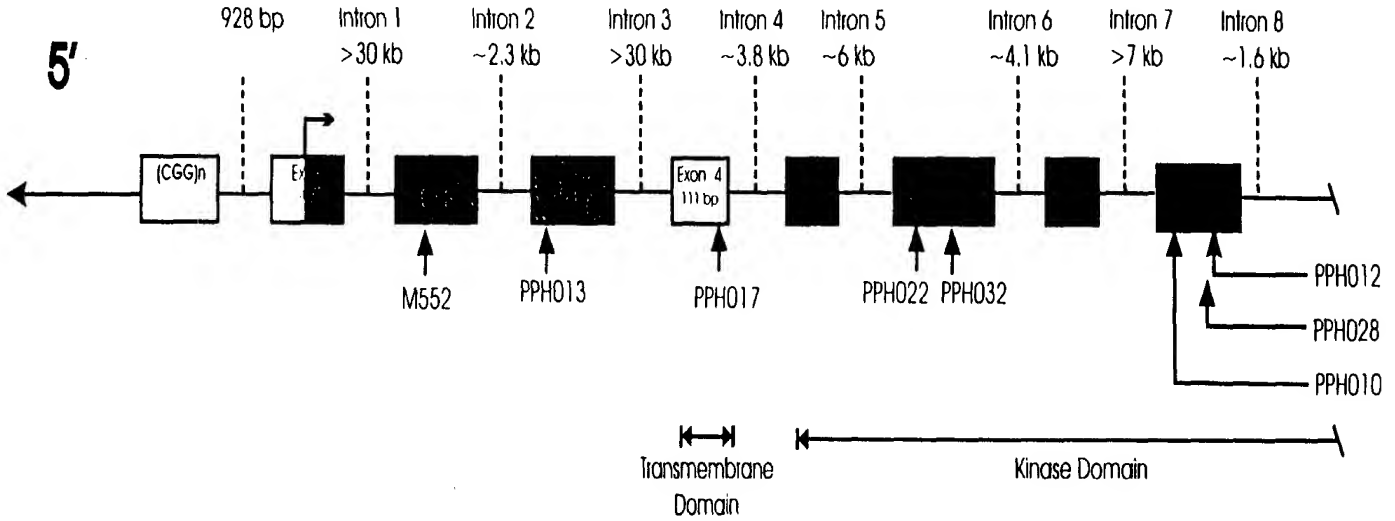
Figure 2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(CGG)n
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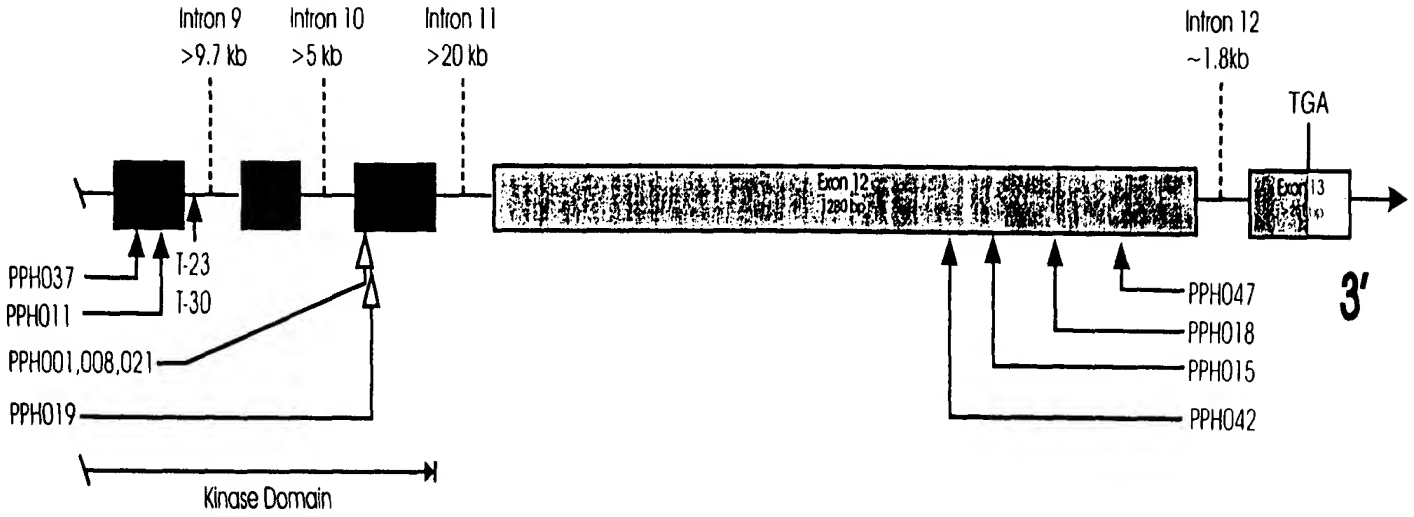
Transmembrane Domain
Kinase Domain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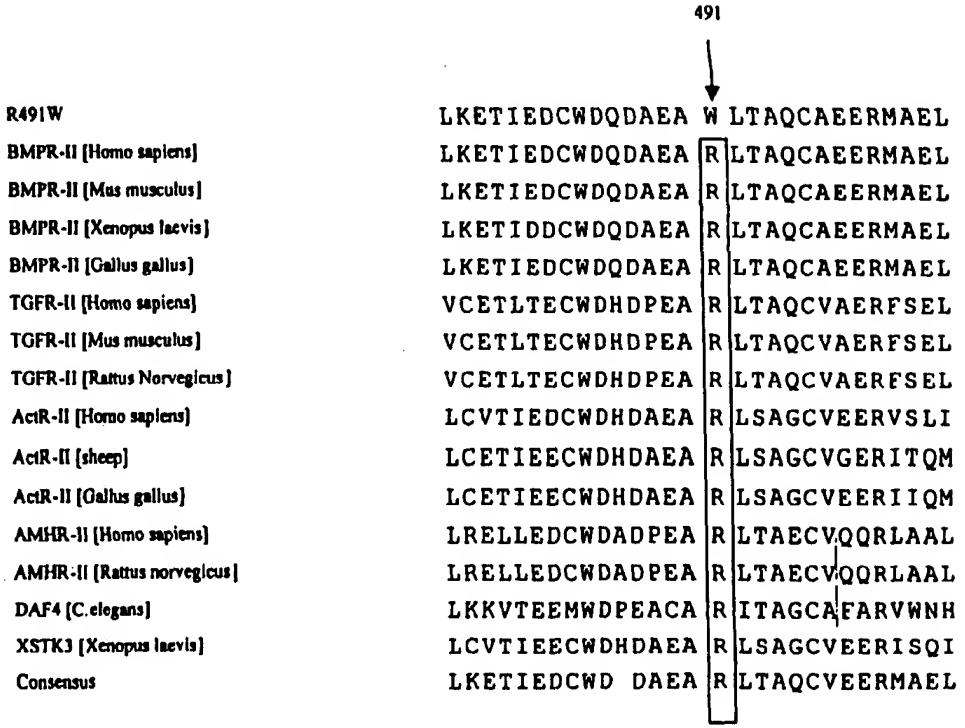
PPH019
PPH047
PPH018
PPH015
PPH042
3′
Kinase Domain

Figure 3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BMPR-II [Xenopus laevis] LKETIDDCWDQDAEA R LTAQCAEERMAEL
BMPR-II [Gallus gallus] LKETIEDCWDQDAEA R LTAQCAEERMAEL
TGFR-II [Homo sapiens] VCETLTECWDHDPEA R LTAQCVAERFSEL
TGFR-II [Mus musculus] VCETLTECWDHDPEA R LTAQCVAERFSEL
TGFR-II [Rattus Norvegicus] VCETLTECWDHDPEA R LTAQCVAERFSEL
ActR-II [Homo sapiens] LCVTIEDCWDHDAEA R LSAGCVEERVSLI
ActR-II [sheep] LCETIEECWDHDAEA R LSAGCVGERITQM
ActR-II [Gallus gallus] LCETIEECWDHDAEA R LSAGCVEERIIQM
AMHR-II [Homo sapiens] LRELLEDCWDADPEA R LTAECVQQRLAAL
AMHR-II [Rattus norvegicus] LRELLEDCWDADPEA R LTAECVQQRLAAL
DAF4 [C.elegans] LKKVTEEMWDPEACA R ITAGCAFARVWNH
XSTK3 [Xenopus laevis] LCVTIEECWDHDAEA R LSAGCVEERISQI
Consensus LKETIEDCWD DAEA R LTAQCVEERMAEL

Figure 4

Family #	#A/#C/#U	Exon #	DNA Sequence Variation	Protein Sequence Variation
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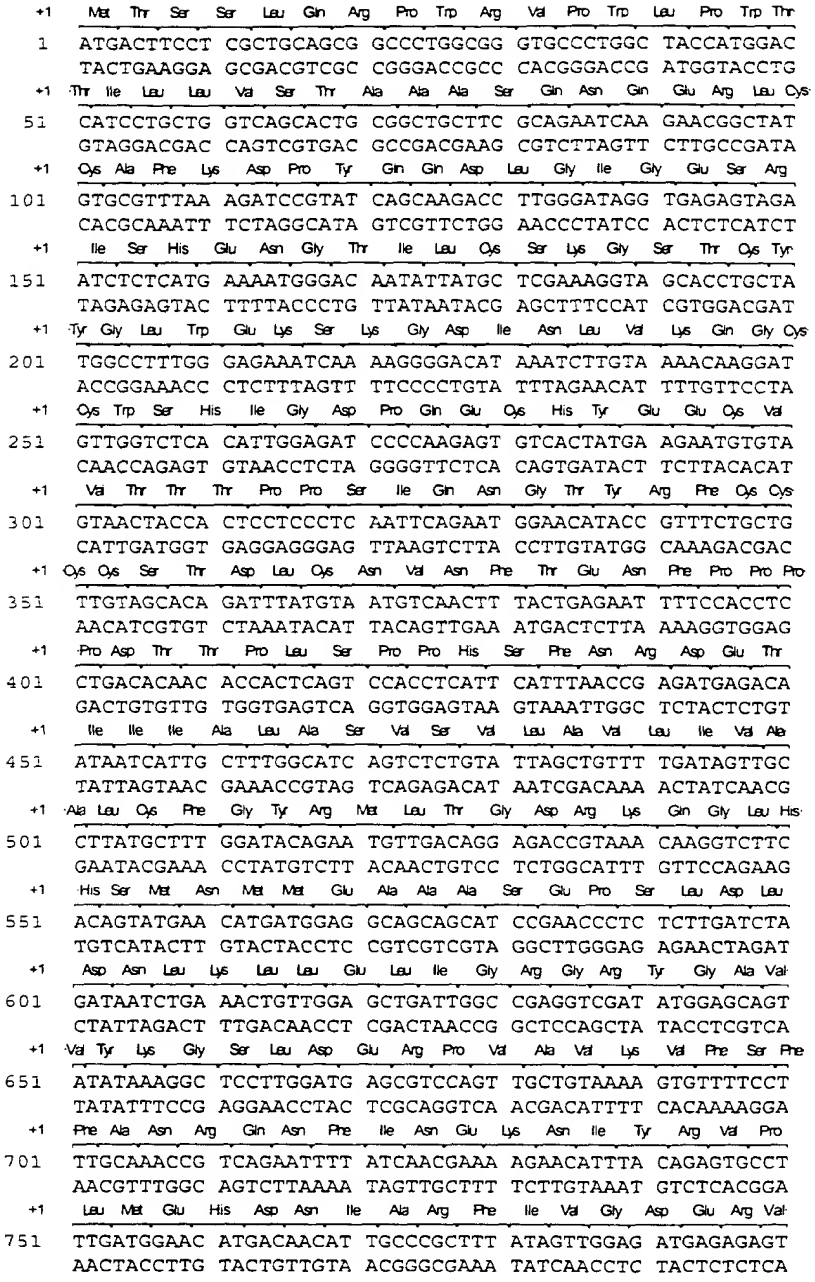
PPH015	6/1/8	12	2579delT	N861fsX10
PPH017	3/0/6	4	507-510delCTTTinsAAA	C169X
PPH018	3/2/4	12	2617C>T	R873X
PPH019	1/0/5*	11	1472G>A	R491Q
PPH022	2/0/0	6	690-691delAGinsT	K230fsX21
PPH011		9	1248-1251delATTT	F417X
PPH012		8	994C>T	R332X
PPH013		3	295T>C	C99R
PPH028		8	1097delG	P366fsX8
PPH032		6	727G>T	E243X
PPH037		9	1214delA	D405fsX6
PPH042		12	2441-2442delAC	H814fsX2
PPH047		12	2695C>T	R899X
M552		2	189-209del21	Del 64-70(STCYGLW)
PPH045		3	296G>A	C99Y
PPH052		3	250T>C	C84R
PPH67-6701		8	1040G>A	C347Y

Figure 5-1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+1 Cys Ala Phe Lys Asp Pro Tyr Gln Gln Asp Leu Gly Ile Gly Glu Ser Arg
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Figure 5-2

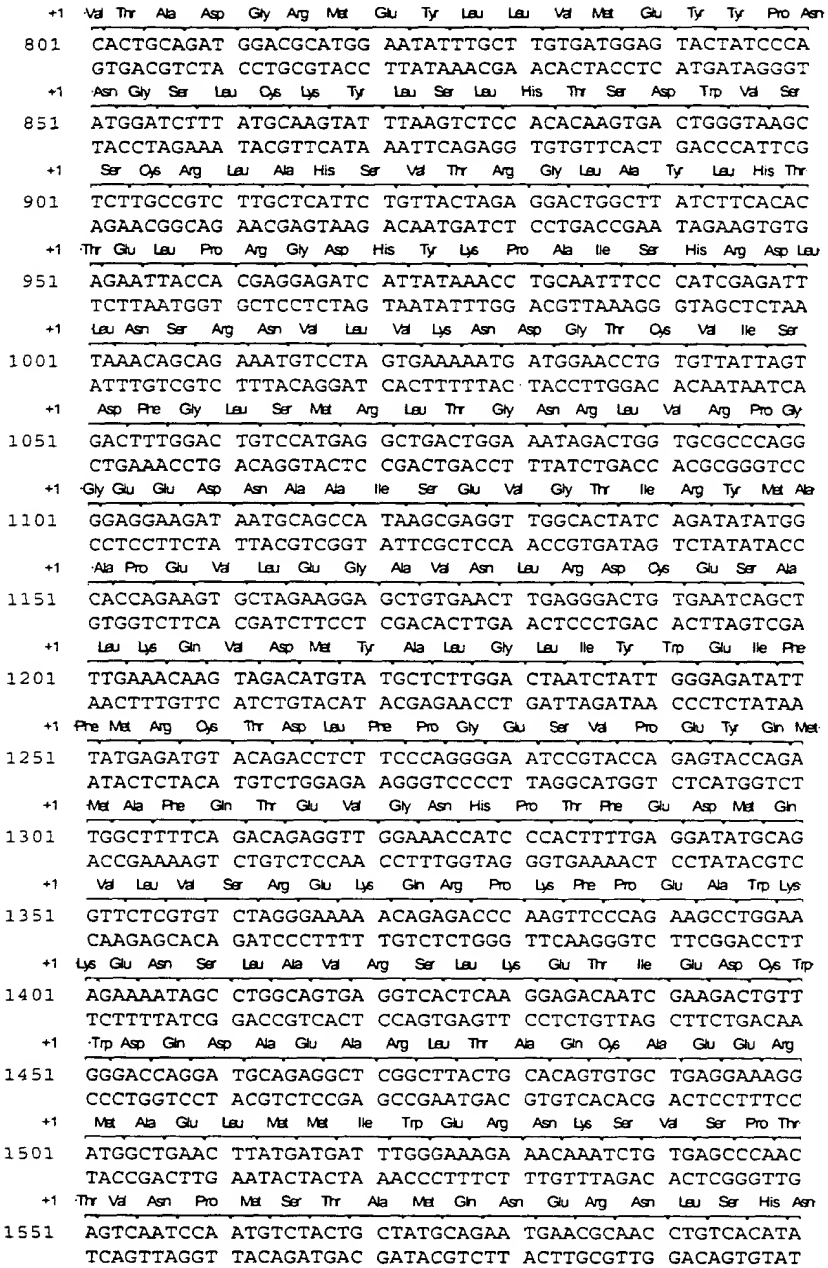
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Figure 5-3

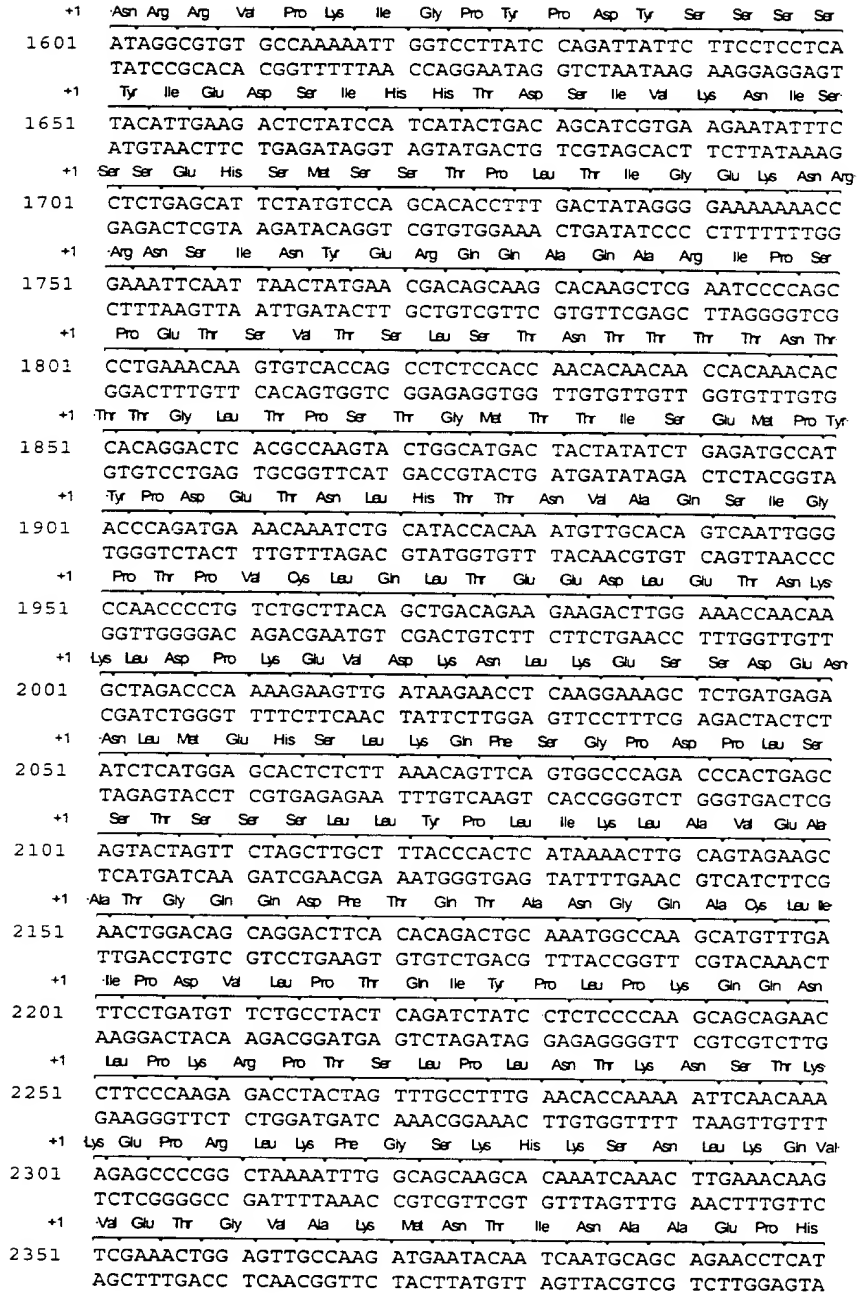
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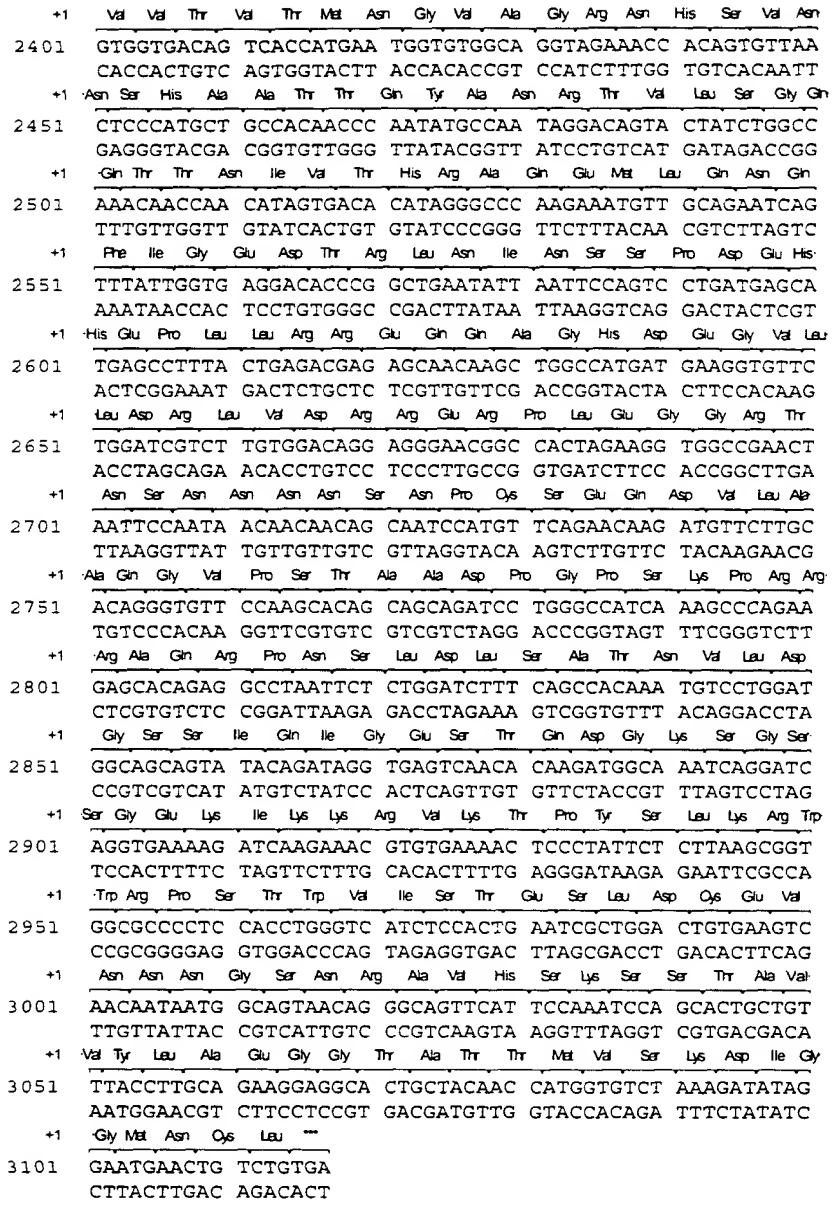
Figure 5-4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